

Genetic Maps → used exclusively as starting point
 HUMAN GENOME PROJECT ← as in sequence of whole genome

POLYGENIC INHERITANCE

★ Many traits not so distinct & spread across a gradient.
 Such traits controlled by 3 or more genes.

Besides involvement of multiple genes, polygenic inheritance takes into account influence of environment

In a polygenic trait, phenotype reflects the contribution of each allele

Human skin colour — polygenic inheritance i.e. effect of each allele additive

All dominant (AABBCC) → Darkest } All recessive (aabbcc) → Lightest

3 dominant allele + 3 recessive allele → intermediate skin colour.

• NO. of each type of alleles in genotype determines darkness/lightness of individual's skin

PLEIOTROPY

★ Single gene → can exhibit multiple phenotypic expression → Pleiotropic gene

• Underlying Mechanism of pleiotropy in most cases effect of a gene on metabolic pathways which contribute towards different phenotypes

• Ex → Phenylketonuria in humans due to (single gene mutation) mutation in genes that codes for enzyme

• Through Phenotypic expression characterised by Phenylalanine hydroxylase
 Mental Retard. Reduction in hair Skin pigment reduction manifests itself

SEX DETERMINATION

★ Experiments on - Insects cytological observations → led to concept of genetic/chromosomal basis of sex determination

★ Henking (1981) traced specific nuclear str. all through spermatogenesis in a few insects → 50% of gamete received it, & 50% did not (sperm. me)
 → could not explain X-body significance. (X-body) ← gave name

actually a chromosome
 X-chromosome

AIR 1747 ↓ sex chromosome

in insects
 XO type → XX-females
 XO-males

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XO → Males — have only 1 X chromosome besides autosomes

→ Females — have a pair of X chromosome " "

Among males, X chr. — present its counterpart → Y chr. = smaller.

In both cases XY, XO type, males produce

either with or without X chr.

Some with X
Some with Y

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male heterogamety

Birds → Female heterogamety

Male - ZZ (Bigger tail)
Female - ZW (Smaller tail)

Some insects

Human Males / Drosophila & Females

Crickshopper

Males

Females

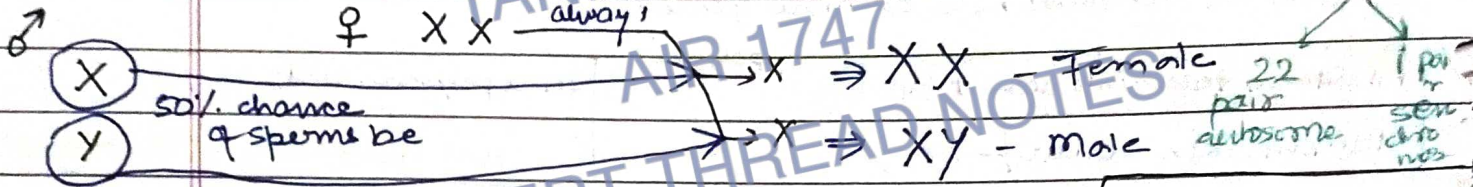
Male XY

Female XX

XO

XX

SEX DETERMINATION IN HUMANS



23 pairs chr.
22 pair autosome
1 pair sex chr. nos

Humans - XY det
ermination of sex

Genetic make up of sperm decides Sex of child.

Each pregnancy - 50% chance of either a male/female child.

SEX DETERMINATION IN HONEY BEE

Sex determination of honey bee based on NO of sets of chromosome an individual receives.

egg + sperm → fertilized egg
develop as → female queen workers.

Unfertilized egg develop as male/drone

By parthenogenesis

Females - Diploid - 32 chromosomes

Males - haploid - 16 chromosomes

Haplo-diploid sex determination system & has special charact. feature

Males → prod. sperm by Mitosis

They don't have father & thus cannot have sons but have grandfather & can have grandsons.

MUTATION

Mutations - results in alteration of DNA sequences & consequently results in

change in genotype & phenotype of org.
Mutation & Recombination Both causes variation in DNA

DNA helix → runs continuously → from one end to other in → Each chromatid (in highly supercoiled form)

Alteration of chr results in Abnormalities/aberration
Since genes located on chr. Alteration results in some of a segment of DNA
Loss (deletion) Gain (insertion duplication) Therefore

(Chromosomal aberrations common in cancer cells (easily observed))

Deletion & Insertion → of bp of DNA causes Frameshift mutation.
Mutation → arise due to change in single bp of DNA
↓ Point mutation

referred to as mutagens. Classic example - Sickle cell anaemia

Chemical Physical factors induce mutation Example → UV rays - can cause mutation

GENETIC DISORDERS

• Pedigree Analysis → After rediscovery of Mendel's work

The practice of analysing inheritance pattern of traits in human being began.

Control crosses → pea plant ✓

In pedigree analysis, the inheritance of a particular trait is represented in family tree over generations.

Human beings X

hence

Study of family history about inheritance of particular trait provides an alternative.

Analysis of traits in several generations of a family

Pedigree analysis

pedigree analysis provides strong tool in human genetics

utilised to trace the inheritance of

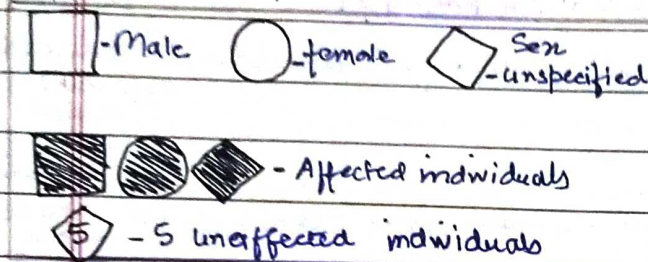
specific trait

Abnormality

Disease

DNA

transmitted from generation to generation without any change/alteration



□ - ○ - mating

□ = ○ - consanguineous mating (mating b/w relatives)

Each & Every feature in organism controlled by one or the other gene located on DNA present on Chromosome

carrier of genetic inf.

MUTATION

However, change or alteration occurs occasionally

Many disorders in human beings have been found to be associated with inheritance of

MENDELIAN DISORDERS

Genetic Disorder → Mendelian disorder
Chromosomal disorder

Determined by → "Alteration / Mutation in the single gene"

These disorders are transmitted to offspring on same lines as we have studied in principle of inheritance

MENDELIAN DISORDERS may be

dominant
Recessive
(Sex linked)

traced by pedigree analysis in a family.

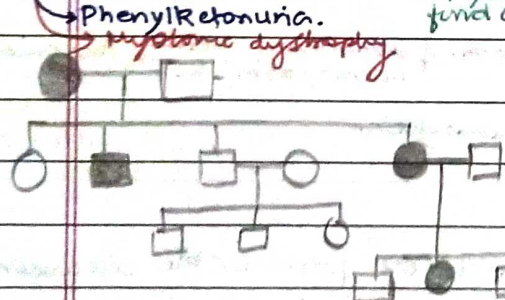
pattern of inheritance of these disorders

- Haemophilia
- Cystic fibrosis
- Sickle cell anaemia
- Colour blindness
- Phenylketonuria
- Myotonic dystrophy

By pedigree analysis we can find out

Ex. haemophilia (X-linked recessive)

Transmission from carrier female to male progeny



Autosomal dominant
- Myotonic Dystrophy



Autosomal recessive
- Sickle cell anaemia

(1) Colour Blindness - Sex-linked recessive due to defect in either red or green cone of eye

Males - 8%

Females - 0.4%

Mutation in certain genes present on X chromosome

this defect is due to

Failure to discriminate b/w red & green colours.

Resulting in

males more → bcz genes that lead to red green colour blindness are on X chromosome. 8 males have only one X while women have 2X.

(2) Haemophilia - Sex linked recessive transmission

due to single protein that is a part of the cascade of proteins involved in the clotting of blood is affected.

hence A simple cut will result in non stop bleeding.

Some of male progeny unaffected carrier female

mother - carrier

- Possibility of female to be haemophilic extremely rare bcz for that Father - haemophilic
- Queen Victoria's Pedigree shows no of haemophilic descendants & carrier of disease
- Unviable in later stage of life

(3) Sickle Cell Anaemia - Autosomal recessive disease

Disease contracted by A single pair of allele \rightarrow Hb^A \rightarrow Hb^S
 $Hb^S Hb^S$ - diseased | $Hb^S Hb^A$ - carrier unaffected (50% possibility of transmitting mutant gene to the progeny)

Both parents heterozygous
 Three genotypes possible \rightarrow $Hb^A Hb^A$ \rightarrow $Hb^A Hb^S$ \rightarrow $Hb^S Hb^S$

Caused by - Substitution of Glutamic acid (Glu) by Valine (Val) at 6th position of β globin chain.
 $GAG \rightarrow GUA$ \leftarrow single base mutation

Mutant Haemoglobin molecule - polymerise under low oxygen tension causing change in shape from biconcave \rightarrow elongated sickle.

Normal Hb(A) gene - Template - GAG -
 Coding - CTC -
 mRNA \rightarrow GAG
 \rightarrow Glu

(Micrograph of RBCs)

Hb^S gene \rightarrow GTC
 - CAC -
 \rightarrow GUA
 \rightarrow Val.

(3) PHENYLKETONURIA - inborn error of metabolism tyrosine

Affected individual - lacks enzyme that

accumulated as phenylpyruvic acid & other deriva tiles

Accumulation is also excreted through urine bcz of its poor absorption by kidney

Mental Retardation causes (accum in brain)

(4) THALASSEMIA: Autosomal-linked recessive

Transmitted when both parents heterozygous.

defect due to Mutation Deletion results in Reduced rate of synthesis of one the globin chains (α - β Ch) \rightarrow causes form. of abnormal haemoglobin \rightarrow results in Anaemia
 classified acc to which chain of haemog. mol. affected. \rightarrow make up haemoglobin

α -Thalassemia	β -Thalassemia	Thalassemia differs from sickle cell anaemia
<ul style="list-style-type: none"> Prod. of α globin chain affected Controlled by 2 closely linked genes \rightarrow $HBA1$ \rightarrow $HBA2$ on chrom. 16 of each parent. Observed due to deletion or mutation of one or more of 4 genes 	<ul style="list-style-type: none"> Prod. of β-globin chain affected. Controlled by - single gene HBB on chr. 11 of each parent. Occurs due to - mutation of one or both the genes 	<p>quantitative \downarrow Synthesising too low/few molecules of globin</p> <p>qualitative \downarrow Synthesising an incorrectly functioning globin</p>

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Chromosomal Disorder



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caused due to 'absence' or 'excess' or 'abnormal arrangement' of 1 or more chromosomes

* Aneuploidy → Gain / Loss of chromosome(s)

(due to) Failure of segregation of chromatids during cell division cycle.

* Down's Syndrome

gain of extra copy of chromosome 21

Turner syndrome

Loss of an 'X-chr.' in human females.

Polyploidy

Failure of cytokinesis after telophase results in ↑ of whole set of chrom

* Total no. of chromosomes in normal human cell

46 (23 pairs)

22 pairs
autosome

1 pair
sex chromosome

Often, seen in PLANTS

TRISOMY of a chromosome

* Sometimes, an additional copy of chromosome

may be

included in an individual

lacking in any individual

MONOSOMY of a chromosome

DOWN'S SYNDROME

(due to) presence of additional copy of chrom. 21
→ Trisomy of 21

first described by Langdon down in 1866

Short statured

Small Round head

Furrowed tongue

Partially open mouth

Broad palm

with characteristic palm crease

Physical
Psychomotor
Mental development retarded.

KLINEFELTER'S SYNDROME

due to additional copy of X-chromosome



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↓ resulting in

Karyotype of 47, XXY

Such individual has

sterile

overall masculine development

Feminine characteristics

development of breast.

Gynecomastia

TURNER'S SYNDROME

due to absence of 1 X-chrom.

↓ resulting in

Karyotype of 45, XO

Such females are

sterile

Rudimentary ovaries

lack of other secondary sexual characters.

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